PAX2 gene

paired box 2

Normal Function

The *PAX2* gene belongs to a family of genes that plays a critical role in the formation of tissues and organs during embryonic development. The members of the PAX gene family are also important for maintaining the normal function of certain cells after birth. To carry out these roles, the PAX genes provide instructions for making proteins that attach to specific areas of DNA and help control the activity (expression) of particular genes. On the basis of this action, PAX proteins are called transcription factors.

During embryonic development, the *PAX2* gene provides instructions for producing a protein that is involved in the formation of the eye, ear, brain and spinal cord (central nervous system), kidney, and genital tract. After birth, the PAX2 protein is thought to protect against cell death during periods of cellular stress.

Health Conditions Related to Genetic Changes

renal coloboma syndrome

More than 20 mutations in the *PAX2* gene have been found to cause renal coloboma syndrome. Most mutations are specific to each affected family; however, one mutation has been found in multiple affected individuals. This mutation inserts one DNA building block (nucleotide) into the *PAX2* gene (written as 619insG). Most mutations occur in the region of the protein that attaches to DNA, impairing its function as a transcription factor. A lack of functional PAX2 protein disrupts the formation of certain tissues (particularly the kidneys and eyes) during embryonic development, causing the signs and symptoms of renal coloboma syndrome.

cancers

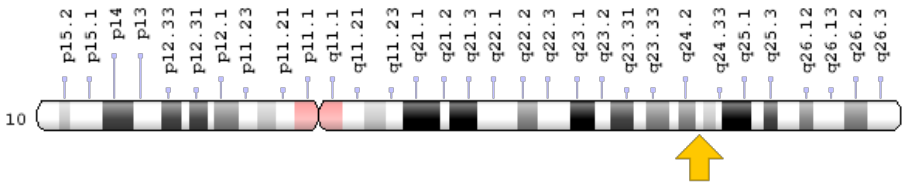
Alterations in the expression of the *PAX2* gene are associated with certain cancers. This altered expression is thought to enhance the gene's ability to protect cells from cell death, allowing for tumor growth (proliferation).

The *PAX2* gene is abnormally active (overexpressed) in certain types of cancer of the kidney, prostate, breast, and ovary. Overexpression of the *PAX2* gene is also seen in a rare form of kidney cancer called Wilms tumor and a soft tissue cancer called Kaposi sarcoma. Kaposi sarcoma causes cancerous lesions to develop under the skin and in mucous membranes (such as the moist lining of the mouth and digestive tract). Typically, high levels of *PAX2* gene expression in cancer cells are associated with a more aggressive cancer.

Chromosomal Location

Cytogenetic Location: 10q24.31, which is the long (q) arm of chromosome 10 at position 24.31

Molecular Location: base pairs 100,732,940 to 100,829,941 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- paired box gene 2
- paired box homeotic gene 2
- paired box protein 2
- PAX2_HUMAN

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Transcription factors https://www.ncbi.nlm.nih.gov/books/NBK10023/#A763
- National Cancer Institute: Kaposi Sarcoma Treatment PDQ https://www.cancer.gov/types/soft-tissue-sarcoma/patient/kaposi-treatment-pdq
- National Cancer Institute: Kidney Cancer https://www.cancer.gov/types/kidney
- National Cancer Institute: Wilms Tumor and Other Childhood Kidney Tumors Treatment PDQ https://www.cancer.gov/types/kidney/patient/wilms-treatment-pdq

GeneReviews

- Renal Coloboma Syndrome https://www.ncbi.nlm.nih.gov/books/NBK1451

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PAX2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- PAIRED BOX GENE 2
  http://omim.org/entry/167409

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/PAX2ID41642ch10q24.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PAX2%5Bgene%5D
- HGNC Gene Family: Paired boxes
  http://www.genenames.org/cgi-bin/genefamilies/set/675
- HGNC Gene Family: PRD class homeoboxes and pseudogenes
  http://www.genenames.org/cgi-bin/genefamilies/set/521
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8616
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5076
- UniProt
  http://www.uniprot.org/uniprot/Q02962

Sources for This Summary

- Amiel J, Audollent S, Joly D, Dureau P, Salomon R, Tellier AL, Augé J, Bouissou F, Antignac C, Gubler MC, Eccles MR, Munnich A, Vekemans M, Lyonnet S, Attié-Bitach T. PAX2 mutations in renal-coloboma syndrome: mutational hotspot and germline mosaicism. Eur J Hum Genet. 2000 Nov;8(11):820-6.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11093271
- Daniel L, Lechevallier E, Giorgi R, Sichez H, Zattara-Cannoni H, Figarella-Branger D, Coulange C. Pax-2 expression in adult renal tumors. Hum Pathol. 2001 Mar;32(3):282-7.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/11274636
- Eccles MR, He S, Legge M, Kumar R, Fox J, Zhou C, French M, Tsai RW. PAX genes in development and disease: the role of PAX2 in urogenital tract development. Int J Dev Biol. 2002; 46(4):535-44. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12141441

- GeneReview: Renal Coloboma Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1451
- Muratovska A, Zhou C, He S, Goodyer P, Eccles MR. Paired-Box genes are frequently expressed in cancer and often required for cancer cell survival. Oncogene. 2003 Sep 11;22(39):7989-97.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12970747
- OMIM: PAIRED BOX GENE 2
  http://omim.org/entry/167409
- Schimmenti LA, Manligas GS, Sieving PA. Optic nerve dysplasia and renal insufficiency in a family with a novel PAX2 mutation, Arg115X: further ophthalmologic delineation of the renal-coloboma syndrome. Ophthalmic Genet. 2003 Dec;24(4):191-202.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14566649

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/PAX2

Reviewed: July 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services